

## Abstracts

# Junior Members Forum

## Thursday 24 November 2005

Ulster Medical Society Rooms, Whitla Medical Building, Belfast



### PROGRAMME

1. 8.00 pm Introduction - Dr Stanley Hawkins, UMS President
2. 8.10 pm Mr Christopher Hoo MRCS (Research Fellow in Surgery) "Molecular Determinants of Prognosis in Malignant Melanoma"
3. 8.25 pm Dr Lorraine Graham MRCP (SpR in Rehabilitation Medicine) "The Physical and Psychological State of Amputees from the Northern Ireland Troubles 1969 – 2003"
4. 8.40 pm Dr Damian McCall MRCP (Research Fellow in Medicine) "Studies on Familial Medullary Thyroid Cancer"
5. 8.55 pm Dr Orla Gray MRCP (SpR in Neurology) "Studies of Mortality in Multiple Sclerosis"
6. 9.15 pm Discussion
7. 9.30 pm Close

### SPOKEN PAPERS

#### S1. Molecular determinants of the invasive potential of malignant melanoma

Chris Hoo, M El-Tanani, FC Campbell.

Department of Surgery, Queen's University Belfast.

**Objective:** The molecular changes associated with transition of melanoma cells to the invasive phenotype are poorly understood.

**Methods:** In this study, B16-F1 melanoma cells will be transfected into a gene construct that may influence cell invasion. Expression of this gene and its coregulators will be assessed by immunohistochemistry in human archival melanoma samples, against the Breslow thickness scale, differentiation and nodal metastasis.

**Outcome:** This study may elucidate molecular determinants of melanoma prognosis and provide novel molecular targets for therapy.

#### S2. A study of the physical rehabilitation and psychological state of patients who sustained limb loss as a result of terrorist activity in Northern Ireland 1969-2003

Lorraine Graham, RC Parke, M Stevenson, M Paterson.

Department of Rehabilitation Medicine, Musgrave Park Hospital, Belfast.

**Objective:** To benchmark the psychological state and physical rehabilitation of patients who have sustained limb loss as a result of terrorist activity in Northern Ireland and to determine their satisfaction with the period of primary prosthetic rehabilitation and the artificial limb.

**Methods:** All patients who sustained limb loss as a result of the Troubles and were referred to our rehabilitation centre were sent a questionnaire. The main outcome measures were the SIGAM mobility grades, the General Health Questionnaire (GHQ12) and 3 screening questions for Post Traumatic Stress Disorder (PTSD).

**Results:** 66% response rate. 52 (69%) patients felt that the period of primary prosthetic rehabilitation was adequate. 32 (54%) lower limb amputees graded themselves SIGAM C or D. 45 (60%) patients stated that they were still having significant stump pain. Significant stump pain and symptoms of PTSD were both associated with poorer mobility. 9 (56%) upper limb amputees used their prosthetic limb in a functional way.

33 (44%) patients showed psychiatric caseness on the GHQ 12 and 50 (67%) had symptoms of PTSD.

**Conclusions:** Most patients felt that the period of physical rehabilitation had been adequate those who did not were more likely to be having ongoing psychological problems. A high percentage of patients continue to have psychological problems and stump pain both of which were associated with poorer mobility.

#### S3. The RET mutation E768D confers a late onset FMTC-only phenotype with incomplete penetrance

Damien McCall, T Dabir,<sup>1</sup> CFJ Russell,<sup>2</sup> PJ Morrison,<sup>1</sup> SJ Hunter.

Regional Centre for Endocrinology and Diabetes and <sup>2</sup>Department of Endocrine Surgery Royal Victoria Hospital, Belfast.

<sup>1</sup>Department of Medical Genetics, Belfast City Hospital.

**Objective:** Mutations of the RET proto-oncogene are associated with MEN and FMTC and aid diagnosis and predictive testing in family members. Genotype-phenotype correlations are also used to plan therapeutic decisions.

**Methods:** We describe a four generation family with a rare E768D mutation in exon 13. The index case was diagnosed with MTC at age 54 and remains free of clinical disease eleven years following thyroidectomy and neck irradiation. Two further family members were identified with MTC at age 25 and 50 years.

**Results:** Of five gene carriers two are asymptomatic at age 70 and 61 years. The former of these asymptomatic carriers has three gene carrier sons who have undergone prophylactic surgery with one having a normal thyroid at age 46, one with C-cell hyperplasia at age 39 and one with a focus of MTC at age 45. No members had evidence of pheochromocytoma or parathyroid disease on screening.

**Conclusions:** The RET E768D mutation is associated with a MTC-only syndrome with a later age at onset, incomplete penetrance and less aggressive clinical course compared with other high risk RET mutations. The appropriate screening strategy for and management of E768D carriers is difficult reflecting the phenotypic heterogeneity.

#### S4. Trends in survival and cause of death in patients with multiple sclerosis in Northern Ireland

Orla M Gray, SA Hawkins

Department of Neurology, Royal Victoria Hospital, Grosvenor Road, Belfast, BT12 6BA.

**Objective:** To investigate trends in survival and causes of death in patients with multiple sclerosis, and to investigate the use of death certification in epidemiological research.

**Methods:** The Northern Ireland Multiple Sclerosis Registry, containing all cases of multiple sclerosis attending neurology outpatients between 1947 and the 1980s, was linked to the Northern Ireland Research and Statistics Agency to identify those who had died and their death certificate documentation.

**Results:** Of 1919 cases on the registry (766 males, 1153 females), 1393 had died, with death certificate data available in 1354 cases, 325 were alive and resident in Northern Ireland and 201 were untraceable. Mean age at onset was 31.16 years and mean age at death 63.65 years. Median survival time was 35 years, with no significant difference between genders, with age of onset or decade of onset. Standardized Mortality Ratios were 1.89 for males (CI 1.73-2.06) and 2.75 for females (CI 2.56-2.95). Multiple sclerosis was documented as the cause of death (Part I) in 27%, as a contributing factor (Part II) in 44% and not at all in 29% of death certificates.

**Conclusions:** Multiple sclerosis is associated with an elevated risk of death with an overall standardized mortality ratio of 2.32. Median survival time was 35 years with no significant difference between genders or with age of onset or decade of onset. Death certificates produce unreliable estimates of mortality rates. In isolation, they underestimate the number of cases and the mean age at death in multiple sclerosis.